Quality of life of patients with Rett syndrome

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Summary

Objectives: Identify in the scientific literature, through the search for scientific articles, aspects of multidisciplinary assistance and the family to promote quality of life for people with Rett syndrome. Methods: review of the literature and the research of qualitative and quantitative approach. Results: in the initial searches were found 150 studies related directly or indirectly with the proposal. Based on reading the titles and developments were excluded 120 publications suited to the goals of the research. Later to do a second analysis with careful reading of these 30 articles selected were excluded more 17, being considered so 13 articles, which had content related to guiding issue. Final considerations: it is essential the participation of the multidisciplinary team, nursing and family, especially in the early years of life, early diagnosis and also the training of professionals in order to meet more appropriately to each specific case. Through monitoring of the multidisciplinary team and the family it was possible to observe that there are improvements in the quality of life of patients with Rett syndrome.

Key words: Rett syndrome, multidisciplinary team, nursing.
**Introduction**

Between the end of the Decade of 1950 and the early 1960, two European doctors, in different situations and circumstances, they began to recognize and become interested in by the clinical picture of a certain condition and behavior that typically affect sex female (who would later be known as Rett syndrome) \(^{(1)}\).

Rett syndrome is a degenerative disease, which was called by that name because the pediatrician Andreas Rett, first described in 1966. However, only had your real disclosure internationally after the publication of the series with 35 cases by Hagberg in 1983.

In 1999, the identification of the mutation of the gene MECP2 (methyl-CpG-binding protein 2) in a large proportion of patients with clinical diagnosis of Rett syndrome confirmed the genetic basis of this syndrome. Previously, your etiology was unknown, and the diagnosis was based on clinical criteria only.

It is understandable that Rett syndrome can be divided into four stages. The first stage, called early stagnation, starting typically between 6 and 18 months of age with a tendency to social isolation, lasting a few months. The second stage, rapidly destructive begins between the first and third year of life and typically lasts from weeks to months. It appears at this stage the psychomotor, reduction imotivado, respiratory irregularities and crying can also be present, epilepsy \(^{(2)}\).

Then, as the third stage, referenced pseudoestacionária, occurs between 2 and 10 years of age having an improvement in some symptoms related directly to the social contact, but in relation to the engine point of view, the presence of ataxia apraxia , spasticity, scoliosis and bruxism. There are frequent episodes of shortness of breath, dyspepsia, forced expulsion of air and saliva.

The fourth step, motor deterioration generally starts late around 10 years and is characterized by a slow progression of engine damage, the occurrence of scoliosis and severe cognitive deviation. And may also occur some neurological disorders.

Girls able to walk independently shall increase the difficulties and in most cases will need a wheelchair.

In the past, the disorder was regarded as a dominant disorder linked to chromosome X, lethal to men, being exclusively observed in women. However, recently, some cases among men have been reported, although with atypical signs and partial syndrome. Today, it is obvious that men can be affected by this condition in some circumstances: boys who have comorbidity with Klinefelter Syndrome, those that present a severe encephalopathy and brothers of affected girls born with losses serious neurological, having usually early death.
Within this context, the research proposed here has your relevance recognized within the nursing and care of the family in relation to the bearers of the Rett syndrome because it is one of the most frequent causes of multiple severe deficiency in females. All of its features, it is a framework that should be of interest to all professionals in the field of health, especially pediatricians, for forwarding and early diagnosis, and specialists who meet people with neuropsychiatric disorders severe.

For professionals in the area of rehabilitation, Rett syndrome is a condition particularly challenging, given the deterioration of motor and cognitive impairment, influencing functional independence level of these patients (3).

The nurse in the therapeutic context is committed to adopting democratic positions, constantly evaluating the proposed activities on assistance, always adapting to the needs of each individual (5).

**Goal**

Considering how important the inclusion of measures that maximize the quality of life of the person with Rett syndrome, this study aims to analyze General through multi-professional assistance procedures aimed at a better life to the patient and how specific objectives identify potential physical, physiological and psychological responses.

**Methods**

It is an integrative review of literature about the improvement in the quality of life of the person with Rett syndrome. This method of research aims to introduce synthetically evidence found in available literature. For preparation we follow some criteria in the following order: definition of the topic and article title, guiding question, criteria for inclusion and exclusion of articles found, including analysis of related to the theme, interpretation of various arguments found and finally, presentation of the review.

The guiding question was: *quality of life of patients with Rett syndrome*. The data were collected in various databases, such as: Latin American literature and Caribbean Health Sciences (Lilacs), Biomedical Literature Citations and Abstracts (PubMed), Medical Literature Analysis and Retrieval System Online (Medline) but the prevailing found in Scientific Eletronic Library Online (Scielo).

The research proceeded in the period from 01 March to 30 June 2016. Inclusion criteria were the articles published by referencing the Rett syndrome and the correlated. Exclusion criteria were already taxes based on the guiding question, thus, the literature found that don't relate to the improvement in the quality of life of disabled people, were discarded.
For data collection, was based on the following information: title of the article, literature database, objectives and timing of references according to the Vancouver style.

**Results**

In the initial searches were found 150 studies related directly or indirectly with the proposal. Based on reading the titles and developments were excluded 124 publications suited to the goals of the research. Later to do a second analysis with careful reading of these selected articles 26, 17 more were excluded, being considered so 09 articles, which had content related to guiding issue.

![Figure 1: search results.](image)

-If the last publications predominated 5 (five) years (14%), followed by studies of the last 2 (two) years (23%), with more than 5 (five) years (63%).

After perusal, Brazilian productions linked to prevailed and therapies that help improve the quality of life.

![Figure 2: Prevalence of publications used.](image)
Discussion

Table 1: scientific articles included in the search.

<table>
<thead>
<tr>
<th>Database</th>
<th>Article title</th>
<th>Main author</th>
<th>Origin</th>
<th>Year of publication</th>
</tr>
</thead>
<tbody>
<tr>
<td>SciELO</td>
<td>1. The neurological expression and genetic diagnosis in Angelman Syndrome, Rett and X-Fragile</td>
<td>Veiga MF</td>
<td>Brazil</td>
<td>2002</td>
</tr>
<tr>
<td>SciELO</td>
<td>2. Role of the psychologist in face of the global child development disorders</td>
<td>Souza JC</td>
<td>Brazil</td>
<td>2004</td>
</tr>
<tr>
<td>SciELO</td>
<td>3. Some considerations on the use of non-traditional therapeutic modalities by the nurse in the psychiatric nursing care</td>
<td>Andrade RLP</td>
<td>Brazil</td>
<td>2005</td>
</tr>
<tr>
<td>SciELO</td>
<td>4. invasive non-developmental Disorders with autistic: Rett syndrome, childhood disintegrative disorder and invasive developmental disorders not otherwise specified</td>
<td>Mercadante MT</td>
<td>Brazil</td>
<td>2006</td>
</tr>
<tr>
<td>SciELO</td>
<td>5. The nurse in the new assistive devices in mental health</td>
<td>Mathew CB</td>
<td>Brazil</td>
<td>2006</td>
</tr>
<tr>
<td>SciELO</td>
<td>7. customer care protocol with respiratory disorder: tool for decision making applied to nursing</td>
<td>Paes GO</td>
<td>Brazil</td>
<td>2014</td>
</tr>
<tr>
<td>SciELO</td>
<td>8. nursing Interventions in Pediatrics: contribution to the measurement of the workload</td>
<td>Ahmed MN</td>
<td>Brazil</td>
<td>2010</td>
</tr>
</tbody>
</table>

Multidisciplinary team and family

Initially, children with Rett syndrome are diagnosed as autistic, due to severe deficiency in social interactions in both syndromes and similarities of disorders in the early stages. However, there are some differences of characteristics that develop in the process of evolution, such as the loss of verbal skills, breathing problems and cranial development slowdown, absent in autism \(^4\).

To enable a better quality of life for the child who is in a deteriorating process, it is necessary the work of psychologist along to teachers that meet the children and the family, accompanying them and clarifying them on the development of syndrome. This treatment may be more effective and have a positive result in
the quality of life of the carrier, when conducted with a multidisciplinary team. Provide welcome to the family, in order to minimize the anguish and suffering, and guide it in dealing with the child's role of the psychologist, who can do it in groups in the case of institutions or individually. When it comes to institutional care, the work of psychologist joins the multidisciplinary team (4).

The bearers of this syndrome can live more than a decade, however, after 10 years of disorder, many patients usually are in wheelchairs (4).

It was observed that there is a strong correlation between self-care and mobility and social function, as well as analyzed that between these areas, functional ability more compromised is the social function soon after that of self-care and finally with best mobility performance (3).

Despite being a progressive neurodegenerative disease, these children respond positively to physical therapy, with new acquisitions. The frequent practice of the March must be one of the goals of physical therapy. When they are under appropriate therapy most children acquires, holds or retrieves the deambulatory skill (1).

The nurse can use music with different purposes and at different times of the patient care: for relaxation, to retrieve memories of past events in the life of the customer. The nurse's role is to analyse at what moment it can use this practice and also evaluate the effects of music on the patient. The psychiatric nurse to use the song for the purposes establishing must associate the like intended carrier not to cause irritation. It appears that she has great power to act on their emotions and behaviors (5).

The role of the psychologist and the multidisciplinary team, in conjunction with the family, resulting in better quality of life for the person with Rett syndrome. Everyone can help build a better day by day and, in particular, the psychologist, who can act directly or indirectly with the feelings, expectations, and desires of a life less painful and more enjoyable (4).

**Nursing**

Nurses deal with uncertainties, even if aware of it. Appropriate decisions requires, in most cases, the articulation of different areas of knowledge that affect human rationality. Evaluate objective data of body or subject by nature subjective is generally not an easy task. Quantify or qualify the uncertainties can assist in the systematization of the care and recovery of the nursing process (6).

The identification and validation of interventions and activities performed by nursing professionals, the use of standardized language to describe the clinical practice of the team, has common meaning where may be communicated in different contexts (7).
The outcome of interventions and activities is a tool that can help in overcoming the difficulties involved in the operationalization of the sizing process nurses\(^{(7)}\).

Among the technologies of nursing care in mental health, the musical intervention contributes directly to the relief of anxiety, stress and to the pursuit of relaxation, besides being used in cases of social isolation. However, despite the recognized beneficial effects, there are few studies on the subject at national level, which may be related to scarce knowledge of music as a therapeutic resource and nursing care element\(^{(6)}\).

It is important that nursing professionals deepen and develop specific knowledge about strategies and methods of the use of music therapy in mental health with the aim of increasing the use your care to children. For that, therefore, more studies are needed and new investigations that contribute to the development and expansion of the use of music as a therapeutic resource in the care in nursing and health\(^{(6)}\). The work of nurses today is the therapeutic agent and the base of this therapy is the direct relationship with the patient and understanding your behaviour. The goal of psychiatric nursing is not the clinical diagnosis or medical intervention, but in fact the commitment with the quality of daily life of the individual in distress\(^{(9)}\).

The nurse must be qualified to act in new ways, with extra hospital services approach and psychosocial rehabilitation. So, seeking the satisfaction of the needs of the patient, assuming various tasks such as greater involvement with the family, adapting to changes arising from mental health policy\(^{(8)}\).

Although the nursing care is based on promotion to life, comfort, dialogue and individual potential of each patient, as the main contributor of the health or disease process, it is necessary to build axles theory and practice in order to improve such assistance. Subsequently, the development and implementation of protocols that meet this perspective, since this is a theoretical-practical support tool, facilitating the planning of care and, consequently, improving the quality of care individual and collective\(^{(6)}\).

The roles of the nurse are also focused on improving mental health, prevention of mental illness, in helping the patient to face the pressures of mental illness and in condition to attend to the patient, the family and the community. For the nurse perform their duties, the observation is very important as well as formulate valid interpretations, the field of action with decision-making, planning assistance, assessing the conduct and the development process. These interventions are part of the nursing process, and should direct the interpersonal skills and therapeutic\(^{(1)}\).

The nurse is more and more present and aware of your new role as a therapeutic agent, keeping in mind that the goal of psychiatric nursing is committed to the quality of life of the individual in suffering, and has ability to exploit several modalities performance of your therapeutic labour activity, putting into practice the patient attention options to consolidate your exercise autonomy and citizenship, or even to
rehabilitate them. These alternatives make the treatment the wearer is less terrible and more pleasurable \(^9\)

**Final considerations**

As Rett syndrome does not cure or specific treatment because the manifestations vary greatly from one patient to another, it is obvious that there are many interventions that can help these patients have a life cycle with less difficulty and healthier. In this way, and the assessment and treatment of those patients are quite complex, it became essential to the multidisciplinary clinical treatment, nursing care process-linked understood as dynamic, innovative, changeable, and also, the active participation of the family.

It was possible to understand the basis of the readings of the studies that the boys may need special attention, for more than the cases prevail in girls, there are situations that the gene MECP2 mutations, for example, individuals with typical karyotype for sex male (46, XY) in children, can take also the neonatal encephalopathy frames and unspecified mental retardation due to your extensive phenotypic spectrum.

Finally, it was possible to observe that in the last 30 years, the knowledge and studies on Rett syndrome have evolved significantly. However, there are still many doctors, therapists and educators who have no idea what really is this syndrome and the vast majority of those who have heard about it, remain without a lot of information about advances in clinical knowledge and used in therapies improving the quality of life, especially in the past decade.

**References**


